

When Baby is a Carrier for Pompe Disease

A small sample of your baby's blood was collected soon after birth and sent to the DHEC Public Health Laboratory for testing. This testing is called Newborn Screening. In SC, newborns are screened for several metabolic and genetic disorders.

Sometimes, a secondary test is needed to help your doctor determine if your baby has one of these disorders. In most cases, the secondary test will be normal. Because an enzyme called Acid Alpha-Glucosidase (GAA) was very low in your baby's first test, a secondary test was performed:

The secondary screening test indicated your baby is a carrier for Pompe Disease

What does it mean to be a **carrier of Pompe disease?**

Carriers of Pompe disease are individuals who have a variation (change) in one of their two GAA genes. These individuals still have one GAA gene without a variation. ***Carriers of Pompe disease do not have any signs or symptoms of Pompe disease.***

However, there is a 1 in 4 (25%) chance a carrier may have a child with Pompe disease if their partner is also a carrier of Pompe. Both parents of a child with Pompe disease are nearly always carriers of the condition.

*Note, babies with a pseudo-deficiency **and** a GAA gene variant most likely do not have Pompe disease either. But baby may need further evaluation with a Metabolic Geneticist to be certain.*

What is Pompe Disease?

Pompe is an inherited condition that affects many different parts of the body. It is considered a lysosomal storage disorder (LSD) because people with Pompe have lysosomes (the recycling centers of each cell) that cannot break down certain types of complex sugars. This causes undigested sugar and other harmful substances to build up in cells throughout the body, resulting in a variety of symptoms.

There are three forms of Pompe Disease (classic infantile onset, non-classic infantile onset, and late onset), which differ in regard to disease severity and age of onset. The symptoms and long-term outcome of each form also vary widely.

Where can I find more information?

Internet References: <http://www.babysfirsttest.org/>